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Dermatoglyphics in Patients With Cenani-Lenz Type Syndactyly: Studies in a New Case

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We describe an additional case of Cenani-Lenz syndactylism in a 4½-year-old boy from a consanguineous Turkish family. The digital anomalies consisted partly of synostosis and partly of malformations of the phalanges. Although there was no radio-ulnar synostosis or abnormality of the bones of the feet, the findings are comparable to those described in the Cenani-Lenz type of syndactyly. We analysed the dermatoglyphics of our patient and compared them with those previously reported. We also investigated the relationship between the bony malformations and the dermatoglyphic patterns in our patient and in the literature. Am. J. Med. Genet. 70:341–345, 1997.

KEY WORDS: syndactyly; synostosis; oligodactyly; hand malformations; dermatoglyphics

INTRODUCTION

The combination of total syndactyly and congenital radioulnar synostosis was first reported by Cenani and Lenz [1967]. It is often associated with mesomelic shortness of the arm, radioulnar and metacarpal synostoses, and disorganized phalanges. The digits are syndactylous, forming a so-called "spoon hand." Radioulnar synostosis varies, and may even be absent; this is also true of abnormalities of the feet.

We present here a boy with Cenani-Lenz syndactyly (CLS). We compare his dermatoglyphics with those of previously published cases and consider the relationship between dermatoglyphics and digital ray maldevelopment.

CLINICAL REPORT

The propositus, SB, a 4½-year-old boy, was referred for evaluation of his malformed hands and feet. He was born following an uneventful pregnancy, the fourth child of a 27-year-old mother and a 29-year-old father. The parents, who were normal, were first cousins, from a small town in South-Eastern Turkey. Three previous pregnancies had resulted in three healthy children (M, F, and F), and the mother had no history of previous abortion or stillbirth. The hands and feet of the parents, and the three sibs, were normal, but it was reported that one of the mother's brothers, also the product of a first cousin marriage, who had died in infancy, had one hand without fingers.

The boy weighed 18 kg (50th centile); he was 107 cm tall (50th centile) and his head circumference was 49 cm. His upper to lower segment ratio was 58/107 (0.6). Other than the limb deformities, there were no significant findings, and he appeared mentally normal. Chest and abdominal X-ray films and intravenous urography were normal.

The hand and foot anomalies were nearly symmetrical (Fig. 1). On each hand there were four grossly abnormal, short, partly syndactylous digits with nails. The left hand had a rudimentary thumb with a nail; the right thumb was completely syndactylous with the second digit, giving the appearance of partial duplication, with a broad bipartite nail having a double face. Because of the partial fixation of the metacarpophalangeal and interphalangeal joints, flexion was functionally restricted. The digits on the left hand were more mobile, permitting the patient to use a pen. Movement of the elbows was not impaired, and there was no mesomelic shortness.

Radiological Findings

Right hand. Three carpals (capitate, hamate, and triquetrum) were present, appropriate for his age. The metacarpals were represented by four bony masses, metacarpals IV and V being almost completely fused. The thumb and second digit showed complete, cutaneous syndactyly. The digit thus formed contained two parallel sets of phalanges, and the distal phalangeal segment had a bipartite nail. The proximal phalanges of those digits deemed to be the third and fourth digits were completely fused and the middle phalanges were articulated by a single joint (see Fig. 2).

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Fig. 1. The appearance of the hands and feet of the patient.

Left hand. Three carpals were present on this hand also. The four metacarpals were unequal in size, and metacarpals IV and V were also fused. The distal end of metacarpal I was narrow and rudimentary. The phalanges were irregularly segmented and partially fused. The second digit had three broad disorganized bones. As in the right hand, the proximal phalanges of those digits deemed to be the third and fourth digits were partially synostosed, forming a fused bony mass, to which the middle phalanges of both these digits were articulated separately. Ulnar ray reduction and partial radial reduction are shown in Figure 2.

Radioulnar synostosis was not present. On examination of the lower limbs, the tibia and fibula were not fused. The talus, calcaneum, and cuboid bones could be recognised separately, appropriate for chronological age, and X-ray studies of the feet showed no gross abnormalities. Both the first metatarsals and their phalanges were broad, with distal lateral deviation and hallux valgus. The second and third toes of both feet showed cutaneous syndactyly.

Dermatoglyphic Findings

The dermatoglyphs of the patient are shown in Figures 3 and 4d.

Right hand. The syndactylous first and second digits showed different types of patterns: radial and ulnar loops. There was a zygodactylous triradius under the partially syndactylous third and fourth digits, and a large whorl at the base of these digits. There was an S-shaped whorl in the thenar area and a distally displaced axial triradius on the palm. However, there was no axial triradius *t,* and the ridges ran transversely on the proximal part of the hand.

Left hand. There was an ulnar loop on the broad second digit of the patient. Over the base of the left syndactylous digits deemed to be the third and fourth digits, there was a zygodactylous triradius peculiar to syndactyly, and there was a large whorl-type pattern beneath these digits. There were no *b* and *c* triradii. There was a whorl (I and I^r loops) within the thenar area. There was a distally displaced triradius (t'') on the left palm, but no proximal axial triradius *t*, and the ridges in the proximal palm were transverse.

The mathematical relationship between the number of loops and triradii [Penrose, 1965] was 7 + 1 = 5 + 3 on the left hand and was 9 (present 7) + 1 = 6 + 4 on the right.

DISCUSSION

Cenani and Lenz [1967] reported on two brothers with a syndrome characterised by a particular type of

Fig. 2. The radiograph of the hands of the patient.

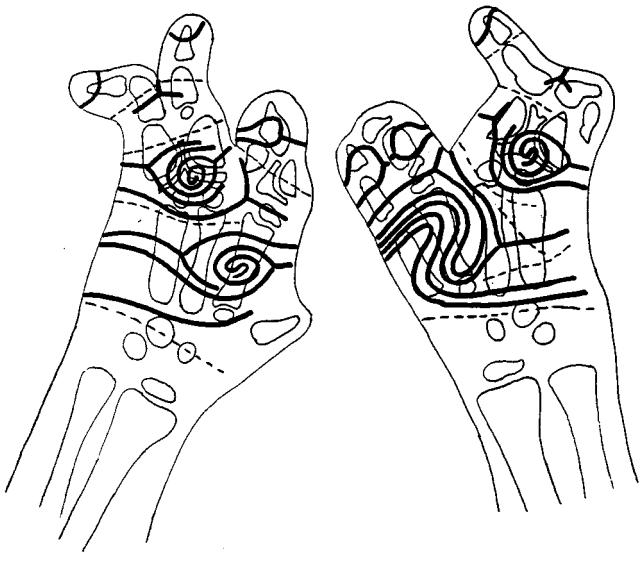

Fig. 3. The dermatoglyphs of the patient superimposed on the radiograph of his hands.

Fig. 4. Comparison of the dermatoglyphic findings in our patient (d) and three other cases previously reported in the literature (a–c; the dermatoglyphics have been derived from tracing of the original palmar photographs). **a**: Cenani-Lenz, 1967, case 1. **b**: Cenani-Lenz, 1967, case 2. **c**: Pfeiffer and Meisel-Stosiek, 1982. **d**: Present case.

syndactyly (seen in Apert syndrome), severe shortness of the radius and ulna, which were also fused, fused metacarpals, and phalangeal maldevelopment. They also investigated patients with similar anomalies previously reported by Liebenam [1938] and Barsky [1958], and a further group of patients reported to date: a pair of twins of discordant sex [Yelton, 1962], a girl [Drohm et al., 1976], a boy with affected siblings [Verma et al., 1976], a brother and sister [Dodinval, 1979], two brothers [Pfeiffer and Meisel-Stosiek, 1982], and a boy [De Smet et al., 1992]. On the basis of their investigations, CLS was classed as an autosomal recessive disorder, an assumption later confirmed by Verma et al. [1976], Dodinval [1979], and Pfeiffer and Meisel-Stosiek [1982]. This also appears to be the mode of inheritance in the family of our patient.

There is considerable variation in the degree of expression of the physical signs in CLS. The feet are generally less severely affected than the hands, and fibulotibial synostosis, which might also be expected, has not been reported to date [Pfeiffer and Meisel-Stosiek, 1982]. Both Dodinval [1979] and Pfeiffer and Meisel-Stosiek [1982] describe patients without radioulnar synostosis, and Pfeiffer and Meisel-Stosiek suggest that these patients constitute a subgroup within CLS. It has been suggested that one way of discriminating between the various different phenotypes found in CLS might be based on an examination of the digital rays, which are often missing or malformed, and we set out to evaluate the use of dermatoglyphics for this purpose in our patient.

The origin of congenital fusions of the limb bones lies in the fifth week of embryonic life, when the limb mesenchyme fails to differentiate into distinct cartilaginous entities, resulting in the union of structures which are normally separate from one another [Dodinval, 1979]. The pattern of abnormalities described by Cenani and Lenz is a variant of a type of homologous malformation of the hands and feet caused by disordered axial and segmentation differentiation, and associated with radioulnar synostosis [see, for example, Verma et al., 1976], and missing or malformed digital rays.

Ridge differentiation begins during the third month of embryonic life, under the stimulus of a growth factor operating between the sixth week and the fourth month of intra-uterine life [Mulvihill and Smith 1969; Schaumann and Alter, 1976]. Malformations of the hands and feet give rise to dermatoglyphic abnormalities at a later stage of development, and these are of

recognisably distinct types in the presence of different types of malformation.

A number of workers [Cummins and Midlo, 1961; Schaumann and Alter, 1976; Loesch, 1983] have described the dermatoglyphic patterns found in syndactyly. Here, transverse ridges cover the palmar areas abutting on the syndactylous digits, which have a zygodactylous triradius at their base. The pattern of ridges may be bilaterally asymmetrical, or one pattern may cover the entire palm. Where digits are missing, the number of digital triradii on the distal palm is reduced, and where there is syndactyly extra digital triradii are present. Where the thumb is absent, the axial triradius *t* is also missing, and the ridges in the proximal palm run transversely [Schaumann and Alter, 1976; Loesch, 1983].

This pattern is repeated in CLS (we have prepared a summary of the findings in our patient, and in some others reported in the literature, see Fig. 4). The hands are grossly abnormal, with large whorls, ulnar and radial loops within the thenar, and interdigital or hypothenar areas of the hands, and these may cover the whole or part of the palm. Cenani and Lenz [1967], in their original paper, found large whorls covering the palms of their two patients with radioulnar synostosis and total syndactyly. The alignment of the ridges on the proximal palm was transverse, and there were no axial triradii (Fig. 4a,b). The patient of Verma et al. [1976], who had radioulnar synostosis, fused metacarpals and metatarsals, ectrodactyly of both hands and one foot, ptosis, nystagmus, and a high-arched palate, also had ulnar loops of the digits. There was a distal loop extending to the ulnar border of the right hand within the thenar area and a radial loop within the hypothenar area of the left hand, extending to the radial side of the hand. This patient had no palmar triradius, but a whorl was present on the left thumb. Dodinval [1979] only described the dermatoglyphics of one of the two patients with oligodactyly and multiple synostoses of the distal limbs. He reported that on the right hand the digital *a* triradius was present, but not the *b, c*, and *d* triradii, and there was no axial triradius *t.* There was an ulnar loop within the thenar area, and a distal loop in the second interdigital area, continuing to an arch over the base of the 3rd digit. The right thumb and 2nd digit showed whorls, and the 3rd digit an arch. On the left hand, the 2nd and 3rd digits were covered in transversely aligned ridges and there was a radial loop on the left thumb. The left palm held *a, a′,* and *d* digital triradii, but no *b* or *c* triradii. There was a distal loop continuing into an ulnar loop between the bases of the 2nd and 3rd digits, and a distal loop near the base of the 3rd digit. There was a loop in the thenar area extending to the ulnar border of the left palm with transverse ridges. Pfeiffer and Meisel-Stosiek [1982] showed a palmar print in a male patient with synostoses of the carpals and metacarpals, and oligodactyly. On the right hand there was a whorl within the second and third interdigital areas, produced by the distal radiant of the *z (ab)* triradius, and a spiral whorl produced by the radial of the axial triradius *t″*. There was a large whorl extending from the radial border to the ulnar side of the distal left palm, formed by the distal radiants of the *a* and *d* triradii, and a large spiral-type whorl within the thenar area, associated with the axial triradius *t″* on the left palm. The alignment of the ridges on the proximal left palm was transverse. Thus, this patient showed distally displaced axial triradii, but neither palm showed a proximal axial triradius (Fig. 4c).

In summary, the dermatoglyphics found in CLS patients reflect their grossly abnormal hand development. There are large whorls, and ulnar and radial loops within the thenar, and interdigital and hypothenar areas of the hands, and these patterns may be asymmetrical, or uniform across the palm, from radial to ulnar side, or in the opposite orientation. The number of digital triradii is reduced and instead zygodactylous triradii are found under syndactylous digits. The axial triradius *t* is absent and the alignment of the ridges of the proximal palm is transverse.

Babler [1987] states that the type of pattern found on the digits is governed by volar pad width at the time of ridge formation, and this in turn is associated with the shape of the bony distal phalanx. Given the extra width of the volar pads found in association with the synostotic bones, this may well explain the large patterns found on the palm of our patient with CLS.

Case 1 of Cenani and Lenz [1967; left palm only], the patient reported by Pfeiffer and Meisel-Stosiek [1982], and our patient do not exhibit proximal axial triradii *t;* there are distally displaced axial triradii (*t‴* or *t″*), and whorls present in the thenar areas, formed by distal radiants of these triradii, and zygodactylous triradii *ab* or *cd* at the base of the syndactylous fingers. There are large whorls at the bases of synostosed digits, and in the thenar areas of the patients' palms. Given their phenotypic similarity, these patients should clearly all belong to the same subclass.

In our patient, due to his syndactyly, it was difficult to determine whether the right thumb was present or absent. According to the dermatoglyphic findings in the proximal palm, the first digital ray, which gives rise to the 1st metacarpal, should be absent, but upon radiological examination the thumb was clearly present, fused with the index finger, although in a hypoplastic state and deviated from its normal position. The 1st digit of the right hand carries a radial loop, which is normally found on the index finger. However, there is an ulnar loop on the adjoining second digit, and for this reason, the first digit should be accepted as the index finger. Further support for this comes from the presence of an axial triradius, although it is not proximally displaced. If the first digit is the index finger, then the 4th and 5th digits are fused. It is interesting to note that if the thumb is displaced, the palmar configurations develop as if it were absent. The dermatoglyphics can be seen superimposed on the bony structure revealed by X-ray examination (see Fig. 3).

The findings in our patient reflect the association between syndactyly and the dermatoglyphic patterns reported in previous cases of CLS, and add to our knowledge of the patterns that may be associated with a malpositioned or aplastic thumb. When examining patients with CLS or other malformations of the hand, the dermatoglyphic patterns should be noted and may

prove to be useful in the description of the condition, although this method cannot take the place of radiological examination.

REFERENCES

Babler WJ (1987): Prenatal development of dermatoglyphic patterns: Associations with epidermal ridge, volar pad and bone morphology. Cell Antropol 11:297–304.

Barsky AJ (1958): "Congenital Anomalies of the Hand and Their Surgical Treatment." Springfield, IL: Charles C. Thomas.

Cenani A, Lenz W (1967): Totale Syndactylie und totale radioulnare Synostose bei zwei Brüdern. Ein Beitrag zur Genetik der Syndactylien. Z Kinderheilk 101:181–190.

Cummins H, Midlo C (1961): "Finger Prints, Palms and Soles." New York: Dover Publications, pp 182–186.

De Smet L, Winnepenninckx B, Fryns JP, Fabry G (1992): Cenani-Lenz type of syndactyly: A complex type of syndactyly with multiple synostoses. Gen Counselling 3:145–147.

Dodinval P (1979): Oligodactyly and multiple synostoses of extremities: Two cases in sibs–A variant of Cenani-Lenz syndactyly. Hum Genet 48:183–189.

Drohm D, Lenz W, Yang TS (1976): Totale Syndactylie mit mesomeler Armverkürzung, radioulnaren und metacarpalen Synostosen und Disorganisation der Phalangen (Cenani Syndactylie). Klin Paediatr 188: 359–365.

Liebenam L (1938): Ueber gleichzeitiges Vorkommen von Gliedmassendefekten und osteosklerotischer Systemerkrankung. Z Mensch Vererbungs Konstitionslehre 21:697–703.

Loesch D (1983): "Quantitative Dermatoglyphics: Classification, genetics and pathology." Oxford, New York, Toronto: Oxford University Press, Part III-8.

Mulvihill JJ, Smith DW (1969): The genesis of dermatoglyphics. J Pediatr 75:579–589.

Penrose LS (1965): Dermatoglyphic topology. Nature 205:544–546.

Pfeiffer RA, Meisel-Stosiek M (1982): Present nosology of the Cenani- Lenz type of syndactyly. Clin Genet 21:74–79.

Schaumann B, Alter M (1976): "Dermatoglyphics in Medical Disorders." New York, Berlin: Springer Verlag.

Verma IC, Joseph R, Bhargava S, Mehta S (1976): Split-hand and split-foot deformity inherited as an autosomal recessive trait. Clin Genet 9:8–14.

Yelton CL (1962): Certain congenital limb deficiencies occurring in twins and half-sibling. Inter-Clinic Inform Bull 1:1–7.